

Management of a pediatric patient with ataxia telangiectasia: Report of a rare case in which diagnostic radiographs are contraindicated

Mebin George Mathew¹

¹Department of Pedodontics and Preventive Dentistry, Saveetha Dental College, Saveetha Institute of Medical and Technical Sciences, Chennai, Tamil Nadu, India

ABSTRACT

Ataxia telangiectasia is a rare neurodegenerative autosomal recessive multisystem disorder which has been reported only once in the dental literature. It is characterized by cerebellar degeneration, telangiectasia, immunodeficiency, and cancer susceptibility and radiation sensitivity. Till date, this disorder remains the only condition in dentistry in which diagnostic radiographs are not allowed. The present case report is aimed to discuss the management of ataxia telangiectasia in a pediatric patient.

Keywords: Ataxia telangiectasia, ataxia telangiectasia mutated, cancer, DNA damage, radiographs

Introduction

Ataxia telangiectasia (AT) is a rare complex progressive disorder characterized by a combination of neurological and systemic symptoms due to the mutation of the ataxia telangiectasia mutated (ATM) gene which impinges on lymphocyte function in AT commonly causing cellular immunodeficiency as well as a predisposition to the development of lymphoid tumors at an early age.^[1,2]

A striking abnormality seen in AT is excessive sensitivity to X-rays as they cause chromosomal breaks.^[2] DNA synthesis resumes quickly following damage which results in faulty DNA synthesis which often results in high number of infections and neoplasms to be associated with AT.^[3] Hence, radiographs should be taken only when necessary.^[1] This case report discusses the management of a pediatric patient with AT.

Address for correspondence: Dr. Mebin George Mathew, Saveetha Dental College, Saveetha Institute of Medical and Technical Sciences, Chennai - 600 077, Tamil Nadu, India.
E-mail: mebingmathew@gmail.com

Received: 12-02-2019

Revised: 28-12-2019

Accepted: 10-01-2020

Published: 28-02-2020

Access this article online

Quick Response Code:



Website:
www.jfmipc.com

DOI:
10.4103/jfmipc.jfmipc_1095_19

Case Report

A 10-year-old boy came to the Department of Pedodontics and Preventive Dentistry with the chief complaint of pain in right and left lower teeth. On examination, occlusal caries was seen with 65 and 85 and grossly decayed 75 was present. Root stumps were seen with respect to 55 and class II cavity with 64 [Figure 1]. Mild gingival inflammation along the accumulation of food debris was seen. The patient had abnormal motor coordination, could not walk without assistance, and had involuntary movements along with ocular telangiectasia [Figure 2]. Family history revealed that he was the second child of consanguineous marriage and his elder sibling was a normal child. The boy could walk normally till he was 6 years old and then developed difficulty to walk. The patient was referred to a pediatrician, where he was diagnosed to be suffering from AT. Consent was received for restorations and extraction but the pediatrician gave precautions for stabilizing the child due to involuntary movements and no diagnostic radiographs were allowed. Restoration of 65 and 85 was carried out with Type II Glass Ionomer Cement (GC Corp, Japan). Extraction was done with 55 and 75 after local anesthesia administration. Patient was

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

For reprints contact: reprints@medknow.com

How to cite this article: Mathew MG. Management of a pediatric patient with ataxia telangiectasia: Report of a rare case in which diagnostic radiographs are contraindicated. J Family Med Prim Care 2020;9:1199-201.

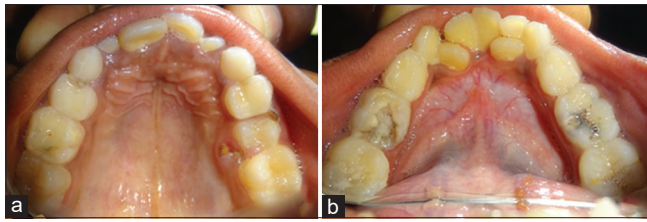


Figure 1: Preoperative photographs. (a) Maxilla; (b) Mandible

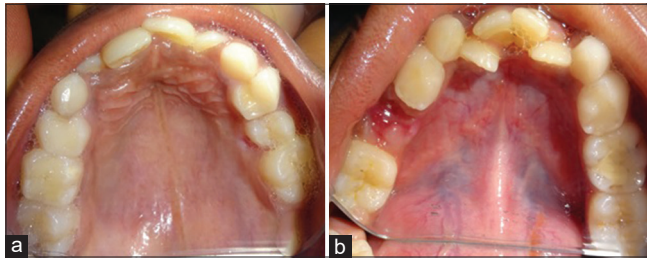


Figure 3: 1 week follow-up. (a) Maxilla; (b) Mandible

recalled after one week and fluoride varnish was applied [Figure 3]. The mother of the patient was advised to brush the teeth of the child twice daily. Oral hygiene instructions were given. The patient returned for a follow-up visit after one year and had maintained good oral hygiene [Figure 4]. Preshedding mobility was seen in 64 and 65. After consultation with the pediatrician, both the teeth were extracted and the patient is still under follow-up.

Discussion

AT was first reported in 1926 with an incidence of 1:100,000 in the general population. The two cardinal conditions required for a patient to be diagnosed with AT are cerebellar ataxia and ocular telangiectasia.^[1,2]

The first symptoms appear in early childhood as the child begins to sit or walk. The child begins to walk normally, but develop an abnormal gait as they grow older and are confined to a wheelchair.^[4] Other characteristics include immunodeficiency, sinopulmonary infection, premature aging, nutritional impairment, dysarthria, and oculomotor abnormalities.^[3,5] An interesting characteristic seen in AT patients is sensitivity to radiation which is often cytotoxic. Hence, X-ray exposure should be limited to times when it is medically necessary for diagnostic purposes.^[1,3,5]

AT is categorized as genomic instability syndrome, disorders that often result in a heightened predisposition to malignancy.^[4,5] Death typically occurs in early or middle adolescence, usually from bronchopulmonary infection, less frequently from malignancy, or from a combination of both.^[1,3] The median age at death is reported to be approximately 25 years.^[1]

A specific treatment to cure AT has not yet been discovered.^[1,3] Since AT presents with a variety of manifestations, a multidisciplinary team is required for management of such patients.^[2] Therefore, addressing the specific symptoms associated with the disease and surveillance to prevent complications is critical.

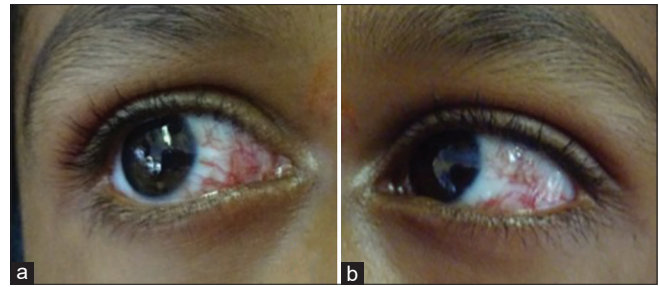


Figure 2: Ocular telangiectasia. (a) Right eye; (b) Left eye

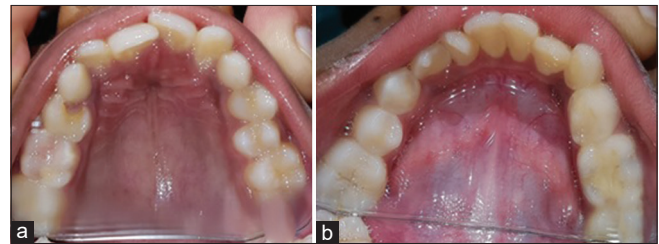


Figure 4: Year follow-up. (a) Maxilla; (b) Mandible

In the present case of the patient, no new carious lesions were seen after one year. This could be due to the oral hygiene instruction given which resulted in good oral health. Though treatment was minimal in this case, the importance lies in the fact that radiographs are contraindicated and can increase susceptibility to infections. Primary care provider should ensure that antenatal diagnosis be performed in families with children affected by AT as pathological mutations of ATM may be found.^[6] Genetic counseling can provide education for families regarding the potential consequences of AT in siblings and other family members.^[7] Hence, practitioners should be aware of this rare disorder and should not take diagnostic radiographs unless necessary for AT patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient's parents have given their consent for the patient's images and other clinical information to be reported in the journal. The patient's parents understand that patient name and initials will not be published and due efforts will be made to conceal patient identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Rothblum-Oviatt C, Wright J, Lefton-Greif MA, McGrath-Morrow SA, Crawford TO, Lederman HM. Ataxia telangiectasia: A review. *Orphanet J Rare Dis* 2016;11:159.
2. Keene EJ. Diagnostic dental radiographs not allowed! A

- child with ataxia telangiectasia. *Eur Arch Paediatr Dent* 2014;15:55-7.
3. Amirifar P, Ranjouri MR, Yazdani R, Abolhassani H, Aghamohammadi A. Ataxia-telangiectasia: A review of clinical features and molecular pathology. *Pediatr Allergy Immunol* 2019;30:277-88.
 4. Duffy R, Liaqat M, Lawrence N, Manders S. Dermatofibrosarcoma protuberans in a pediatric patient with ataxia telangiectasia syndrome. *Pediatr Dermatol* 2019;36:400-1.
 5. Mei L, Zhang J, He K, Zhang J. Ataxia telangiectasia and Rad3-related inhibitors and cancer therapy: Where we stand. *J Hematol Oncol* 2019;12:43.
 6. Fiévet A, Bellanger D, Rieunier G, Dubois d'Enghien C, Sophie J, Calvas P, *et al.* Functional classification of ATM variants in ataxia-telangiectasia patients. *Hum Mutat* 2019;40:1713-30.
 7. Schon K, van Os NJH, Oscroft N, Baxendale H, Scoffings D, Ray J, *et al.* Genotype, extrapyramidal features, and severity of variant ataxia-telangiectasia. *Ann Neurol* 2019;85:170-80.